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ABSTRACT

A review of the research literature related to Williams 'elfin facies' Syndrome (a condition including mental retardation and cardiac anomalies), a case study, and the results of an early intervention program, are reported. The medical aspects of the syndrome have been reported with sufficient consistency for a profile to emerge. Psychological variables have been reported with less frequency and educational programing needs have received little attention. Etiology and prognosis have been considered indeterminate. The case study reported a 27 month intervention program (9 months to 36 months) and included an abbreviated history, comparison of case to syndrome profile and the results of educational programing. Recommendations for appropriate educational programing are made in light of previous research and results of the reported case.
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Williams 'elfin facies' Syndrome:
A Case Study

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Williams 'elfin facies' Syndrome is a congenital structural malformation generally characterized by a distinctive and peculiar facies, mental retardation, infantile hypercalcemia and supra-ventricular aortic stenosis. Etiology and prognosis of this syndrome are indeterminate. A review of the pertinent literature, and a case study with emphasis on psychological profile and the results of early educational programming are presented in this paper.

REVIEW OF THE LITERATURE AND CLINICAL FEATURES

Review of the Literature

Williams, Barratt-Boyes and Lowe (1961) suggested that a close facial resemblance was diagnostic of a new syndrome that included mental retardation and supra-ventricular aortic stenosis. Black and Bonham Carter (1963) noted similar peculiar facies of children with idiopathic hypercalcemia of infancy with failure to thrive (Lightwood, 1932) and supra-ventricular aortic stenosis and suggested that children with the cardiac anomalies were survivors of the infantile hypercalcemia syndrome. Jones and Smith (1975) attempted to establish a clear diagnostic profile using the characteristics frequently reported in the literature in the major areas of growth: central nervous system function, facies, limbs and cardiovascular anomalies. No characteristics were reported for all subjects (N=19); however, 75 percent or more were reported as exhibiting prenatal and postnatal growth deficiencies, mental deficiency, medial eyebrow flair, prominent lip with open mouth, displacement of the large toe toward other toes, heart murmur and hoarse voice. The syndrome is somewhat inconsistent in clinical features, a consideration important to correct diagnosis and appropriate prognosis.

Clinical Features

Peculiar facies. The elfin facies is apparent at birth and becomes increasingly distinctive with maturation. General agreement continues with the original description reported by Williams, et al., (1961) that included a broad forehead, wide-set eyes,

heavy cheeks; a mouth which is wide and pouting, pointed chin, large ears, blue irises and malocclusion of the teeth (Satran and Fisch, 1977; White, Preus, Watters, and Fraser, 1977; Jones and Smith, 1975; Copperman and Low, 1975; Losekoot and Nelis, 1975; Kelley and Barr, 1975; Hoeffel, Henry, Jimenez, and Pernot, 1974; Beuren, Schulze, Eberle, Harmjanz, and Apitz, 1964; Beuren, Apitz, and Harmjanz, 1962).

Though all of the peculiar facies characteristics are not exhibited by each subject, they are generally found in sufficient number and combination to make the elfin-like appearance readily recognizable.

Mental retardation. Jones and Smith (1975) reported mental deficiency as one of the most consistent distinguishing features of the syndrome (94 percent of their sample). The presence of mental retardation in the syndrome is consistently reported in the literature (Bennett, LaVeck, and Sells, 1978; Daiger, Miller, Romeo, Parsons, and Cavalli-Sforza, 1978; Neilson and Hossack, 1978; Satran and Fisch, 1977; Folger, 1977; White, et al., 1977; Jensen, Warburg, and DuPont, 1976; Becroft and Chambers, 1976; Jones and Smith, 1975; Mehes, Szelid, and Toth, 1975; Copperman and Low, 1975; Losekoot and Nelis, 1975; Battle, Harris, Pashayan, and Pruzansky, 1975; Kelly and Barr, 1975; Usher, Goulden, and Murgo, 1974; Hoeffel, et al., 1974; Forbes, Bryson, Manning, Amirhakimi, and Reina, 1972; Wiltse, Goldbloom, Antia, Ottesen, Rowe, and Cooke, 1966; Beuren, et al., 1964; Garcia, Friedman, Kaback, and Rowe, 1964; Black and Bonham Carter, 1963; Beuren, Apitz, and Harmjanz, 1962; Williams, et al., 1961).

Intellectual functioning is generally reported in the mild to moderate range of retardation though some cases of severe retardation have been reported (Hoeffel, et al., 1974; Wiltse, et al., 1966).

Bennett, et al., (1978) used the McCarthy Scales of Children's Abilities to compare the skill patterns of seven children diagnosed as Williams 'elfin facies' Syndrome to the skill patterns of nine control children exhibiting developmental problems. Mean scores differed significantly on the subscales for motor and perceptual

performance, while there was no significant differences on the verbal memory and quantitative subscales. The developmental profile for the Williams 'elfin facies' children showed a distinct pattern; highest scores were attained on the three language subscales while scores for the motor and perceptual performance subscales were noticeably depressed. Cognitive development appeared to progress at an even rate as exhibited in measurement of abilities at intervals of up to five years.

Hypercalcemia of infancy and failure to thrive. Clinical symptoms of hypercalcemia include some combination of the following: weight loss or inability to gain weight, constipation, diarrhea, constipation alternating with diarrhea, vomiting, cough, restlessness, muscular hypotonia, anorexia, and cyanosis. Chronic renal disease is often present as well as the cardiac signs of systolic murmur and occasional hypertension. Serum calcium is usually elevated and osteosclerosis and microcephaly have been reported as skeletal findings. Clinical signs of hypercalcemia invariably begin during the first year of life (Jensen, et al., 1976; Jones and Smith, 1975; Battle, et al., 1975; Kelly and Barr, 1975; Wiltse, et al., 1966; Black and Bonham Carter, 1963; Forfar, Tompsett, and Forshall, 1959; Fellers and Schwartz, 1958; Forfar, Balf, Maxwell, and Tompsett, 1956; Bonham Carter, Dent, Fowler, and Harper, 1955).

Cardiac anomalies. Supravalvular aortic stenosis is generally regarded as a common characteristic of children with Williams 'elfin facies' Syndrome (Folger, 1977; Becroft and Chambers, 1976; Friedman and Mills, 1969; Beuren, et al., 1962; Williams, et al., 1961) although it is not present in all cases (White, et al., 1977; Wiltse, et al., 1966). Jones and Smith (1975) report that only 32 percent of Williams 'elfin facies' patients they examined exhibited supravalvular aortic stenosis and conclude that it is not a necessary component of the syndrome.

Peripheral pulmonary stenosis has been identified as an additional cardiac malfunction characteristic of this syndrome (Copperman and Low, 1975; Wiltse, et al., 1966). That a distinct relationship in cardiac etiology for cases exhibiting both

supravalvular aortic stenosis and peripheral pulmonary stenosis has also been suggested (Beuren, et al., 1964). This relationship is noteworthy in regard to prognosis because peripheral pulmonary stenosis is presently considered inoperable. Cardiac anomalies of the type characteristically associated with Williams 'elfin facies' Syndrome are suggested by murmurs, dyspnea, or failure to thrive.

Dental anomalies. Beuren, et al., (1964) described dental malformations as one of the distinguishing characteristics of the syndrome they labeled supravalvular aortic stenosis. The dental anomalies of the ten patients reported included small, hypoplastic teeth, bud-shaped teeth, incomplete dentition for both deciduous and permanent teeth, dysgnathism, partial adontia, and delayed mineralization. They conclude that the dental malformations partially cause the 'elfin facies' which is characteristic of this syndrome. Their findings have been confirmed by other researchers (Kelly and Barr, 1975; Battle, et al., 1975; Friedman and Mills, 1969) leading to a general acceptance of dental anomalies as an additional major characteristic of the syndrome.

Psychological variables. Williams children have been variously described as friendly, outgoing, charming, lovable, pleasant, overly affectionate, loquacious, talkative and behaving in a cocktail party manner (Bennett, et al., 1978; Neilson and Hossack, 1978; Folger, 1977; Jones and Smith, 1975; Usher, et al., 1974; Forbes, et al., 1972; Beuren, et al., 1964; Beuren, et al., 1962). Beuren, et al., (1964) provided a "typical" description of the type of personality exhibited in this syndrome: "All have the same friendly temperament. They are very active and always happy. The similarity of appearance is even more striking when they talk, all with the same deep somewhat metallic voice. In total they are what one would describe as charming characters." (p. 472).

The assumption that most children with Williams 'elfin facies' Syndrome have charming personalities is certainly an oversimplification of their personality patterns. In contrast to the early descriptions, behavior problems are often severe,

including the descriptors negative, impulsive, self-destructive and hyperactive (Jones and Smith, 1975; Usher, et al., 1974).

Etiology

The etiology of Williams 'elfin facies' Syndrome is unknown. Excessive intake of vitamin D inutero or a genetically determined sensitivity within the child to normal levels of vitamin D has been suggested as a cause of infantile hypercalcemia. However, this cannot be demonstrated in all cases; and all children receiving high doses of vitamin D did not develop hypercalcemia (Daiger, Miller, Romeo, Parsons, and Cavalli-Sforza, 1978; Folger, 1977; Becroft and Chambers, 1976; Jones and Smith, 1975; Friedman and Mills, 1969; Forfar, Tompsett, and Forshall, 1959; Fellers and Schwartz, 1958; Forfar, Balf, Maxwell, and Tompsett, 1956; Lowe, Henderson, Park and McGreal, 1954). Chromosome anomalies have not been reported, as was expected because of a typical syndrome facies similar to that found in Down's Syndrome (Beuren, et al., 1962; Beuren, et al., 1964).

The possibility of a genetic origin has been reported by some (Neilson and Hossack, 1978; Satran and Fisch, 1977; White, Preus, Watters, and Fraser, 1977; Becroft and Chambers, 1976; Morrison and McNally, 1967; Merritt, Palmer, Lurie, and Petry, 1963) and rejected by others (Jones and Smith, 1975; Beuren, et al., 1964). Research evidence cannot be considered conclusive at this time.

CASE STUDY

Developmental History

J.P. was born naturally after an uneventful pregnancy. Labor had an approximate duration of 10 hours. There was no utilization of an anesthetic or forceps and the mother was awake when the baby was born, head first. J.P.'s Apgar scores at birth were 9 and 10. She was small for her gestational age, weighing 5 pounds 11 ounces and measuring 19½ inches in length. At birth, she exhibited an abnormal facies with prominent eyes, particularly evident when crying. She was born with jaundice and only one umbilical artery.

At the time of her discharge she had a heart murmur at the left sternal border which progressed from grade II/VI to grade IV/VI over a two-week period of time. In addition, she exhibited congestive heart failure during her two-week check-up. A cardiac catheterization when she was six-months old showed coarctation of the aorta with peripheral pulmonary artery stenosis and also possible sub-pulmonary and sub-aortic stenosis with generalized hypoplasia for the arterial system.

Initially, J.P. was breast fed but she was weaned at nine weeks. It was reported that she ate poorly and had difficulty gaining weight, although she had no reported difficulties with vomiting, colic, constipation, or diarrhea. At eight and one-half months she weighed 12 pounds and 4 ounces and was 25 3/4 inches long. She slept only eight to nine hours per night in her own room and was described as a thin, unhappy infant. She could not crawl or sit at that time, but was capable of rolling over. The clinical diagnosis - complex congenital heart disease with a coarctation of the aorta with hypoplastic isthmus, hypoplastic arterial system, both right ventricular and left ventricular outflow obstruction probably secondary to hyperdynamic septum, peripheral pulmonary artery stenosis, and possible cardiomyopathy.

A chromosome analysis showed a normal 46 XX karyotype. Her growth curve was delayed but parallel to normal. In addition, her left pupil was reported to be slightly larger than the right, she had a stellate pattern in her irises, blue eyes, recurrent otitis media in both ears, hips which were difficult to abduct, stubby fingers, mild clinodactyly of the fifth fingers, and a bilateral valgus positioning of the large toe. Her facial characteristics were described as including a depressed nasal bridge, a medial eyebrow flair, pre-orbital fullness, somewhat prominent lips, and anteverted nares.

At 17 months, J.P. was reported to be physically developing below the normal levels for her age; height was at the 10th percentile while weight and head



PLATE 1. Williams 'elfin facies'
Syndrome, female, age 30 months



PLATE 2. Peculiar facies; full
cheeks, depressed nasal bridge,
short nose, pointed chin



PLATE 3. Excessive abdominal protrusion and slight limbs symptomatic of cardiac condition



PLATE 4. Oral configuration; small hypoplastic teeth, bud-shaped teeth, malocclusion

circumference were both below the 5th percentile. The diagnosis was altered to Williams 'elfin facies' Syndrome.

Between 18 and 36 months, J.P. continued to physically develop below normal levels; with height and weight remaining in the 10th percentile. The cardiac anomalies were determined to be operable but could not be scheduled until body weight increased. Dental anomalies noted included hypoplasticity, bud-shaped teeth and malocclusion. The peculiar facies was judged to be increasingly pronounced.

Psychological Profile

The Bayley Scales of Infant Development (Bayley, 1969) was administered at 18 and 30 months; 18 month scores, Mental Development Index 70, Psychomotor Development Index 56, 30 month scores, Mental Development Index 92, Psychomotor Development Index 88. These increases are considered significant and attributed, in great part, to the intervention program described later.

Adjudged temperament for J.P., though clearly within normal limits, cannot be considered overly pleasant or outgoing as reported in various studies. Using a combination of the Bayley Infant Behavior Record and clinical judgement, J.P. was considered to lag to a large degree in social skills up to age 18 months and to a lesser degree on emotional variables. Considerable growth was noted with increased age, appropriate social contacts and increased parenting skills. J.P. continued to exhibit defiant behaviors and temper tantrums throughout the investigation period.

Intellectual functioning exceeds the generally expected prognosis for Williams 'elfin facies' Syndrome. J.P. continues to function in the borderline normal range. Personality characteristics associated with this syndrome are absent with a substantially normal repertoire of age-appropriate behaviors exhibited.

Educational Program

J.P. was referred to the University 0-3 Handicapped Program at nine months of

age. A program was developed that targeted gross motor, fine motor and sensory motor developmental goals. Services were provided by a staff occupational therapist and a child development specialist who worked directly with the child and with the parents to increase basic stimulation and developmental activities. All programming between ages 9 months and 18 months was provided in the home.

At age 18 months, J.P. was enrolled at the University Infant Center and a program was designed that provided daily individual instruction under the supervision of Department of Special Education and University Speech and Hearing Clinic faculty. Program goals were designed to coincide with the Guide to Early Developmental Training (Wabash Center, 1972) which was used as an activity guide and a progress monitor. The areas of language, perceptual motor, locomotion, body image and balance and posture were specifically targeted. A 30-minute video-taped behavior sample was made every two weeks that monitored pupil progress, teacher/pupil interaction and established a data base that could be objectively analyzed. This program was discontinued at age 36 months when J.P. was enrolled in the preschool handicapped class.

The Receptive-Expressive Emergent Language Scale (Bzoch and League, 1971) was administered at program entrance and again at 12, 24 and 36 months.

	10 Mos.	12 Mos.	24 Mos.	36 Mos.
Receptive Language Age	4 Mos.	12 Mos.	16 Mos.	24 Mos.
Expressive Language Age	6 Mos.	12 Mos.	18 Mos.	21 Mos.
Combined Language Age	5 Mos.	12 Mos.	17 Mos.	22.5 Mos.

Attainment of goals was reported for the checklist accompanying the Guide to Early Developmental Training. Skill area and percent of attainment reached within normal age limits can be summarized as follows:

Area	% - 24 Mos.	% - 36 Mos.
Language	42	68
Perceptual Motor	45	51
Locomotion	26	54
Body Image	16	66
Balance and Posture	38	60

J.P. was screened at 24 and 36 months using the Denver Developmental Screening Test (Frankenburg, et al., 1967) simultaneously with other preschool-handicapped children. Results on the 24-months screening showed three delays in the personal-social area, one in the fine-motor adaptive, no delays in language, and four in gross motor. Results on the 36-months screening showed four delays in personal-social (three continued from 24 months), none in fine-motor adaptive, none in language, and three in gross motor (two continued from 24 months).

Though continued delay was exhibited, growth was found to parallel normal developmental milestones. All assessment confirmed language strengths and motor deficits.

Discussion

Considerable literature details the medical aspects of Williams 'elfin facies' Syndrome. Some degree of mental retardation is reported with considerable frequency. There is a clear need for studies that examine psychological variables and the results of educational programming. Systematic differential diagnosis should begin with 'elfin facies', high probability of developmental delay, suspected cardiac anomalies and expand to include secondary syndrome characteristics. Prognosis will continue to be speculative until data is accumulated that reports educational programming results. The clinical picture of Williams 'elfin facies' Syndrome remains incomplete.

The case reported was monitored in a 27-month educational program. Though some delay continued, the growth was considered sufficient to establish considerable efficacy for early diagnosis and special education intervention. Strengths in language and weaknesses in motor areas were noted and considerable growth in intellectual functioning was evidenced. Personality characteristics were not consistent with much of the literature reviewed, to the extent that the inclusion of behavior typical of the syndrome might need reconsideration. There is sufficient evidence in the research literature and confirmed in this case to suggest that a

Williams 'elfin facies' Syndrome diagnosis by medical personnel should generate an early referral for psychological evaluation and educational programming.

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